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Stüve-Wiedemann Dysplasia in a 3½-Year-Old Boy

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Stüve-Wiedemann osteochondrodysplasia is a rare disorder with distinct clinical and diagnostic radiographic findings. The condition is classified as a bent-bone dysplasia with early, lethal outcome. We report on a boy with Stüve-Wiedemann syndrome who is well and alive at the age of 3½ years.

KEY WORDS: campomelic, kyphomelic, Stüve-Wiedemann

INTRODUCTION

There are three well-differentiated bone dysplasias which share bowed long bones as the most important clinical and radiographic abnormality. The most common, usually with early fatal outcome is campomelic dysplasia [Reichenbach et al., 1994]. Kyphomelic dysplasia is much rarer [Temple et al., 1989]. This disorder usually has a good prognosis. Stüve-Wiedemann dysplasia is the rarest and least well known [Stüve and Wiedemann., 1971]. We report on a boy with Stüve-Wiedemann dysplasia who survived a turbulent infancy and is well at 3½ years.

CLINICAL REPORT

This boy was the only child of non-consanguineous 26-year-old Caucasian parents. There was no family history of malformations. The pregnancy was uneventful,except for respiratory infections at 12 and 16 weeks treated with paracetamol and traumatic perforation of the eardrum at 5 months treated with amoxicillin and clavulanic acid. Fetal movements began at 5 months of gestation and were considered "feeble". Ultrasound findings at 4, 5, and 6 months of gestation were reported as normal.

The delivery at gestational age of 40 weeks was normal. Birth weight was 3,760 g (50–70th centile), length as 51 cm (50th centile), and head circumference was 36.5 cm (75–90th centile). Apgar scores were 6 and 9 at 1 and 5 minutes. Distal arthrogryposis was diagnosed at birth. Between the 3rd and 14th week of life he had recurrent attacks of fever up to 39°C every week. At 7 months he was re-admitted with a fever of 39.6°C. Episodes of pyrexia up to 38°C persisted until the age of 24 months. The ESR and leucocyte count were elevated. The boy also had severe sucking and swallowing difficulties necessitating tube feeding until age 8 months.

Extensive hormonal,metabolic and immunological studies showed no abnormality. Bacteriological and viral studies were noncontributory. Cause of the hyperthermia was never established.

At 18 months his weight and length were between the 3rd and 10th centiles. Some shortness of the limbs was noted at that time and a bone dysplasia was sus-

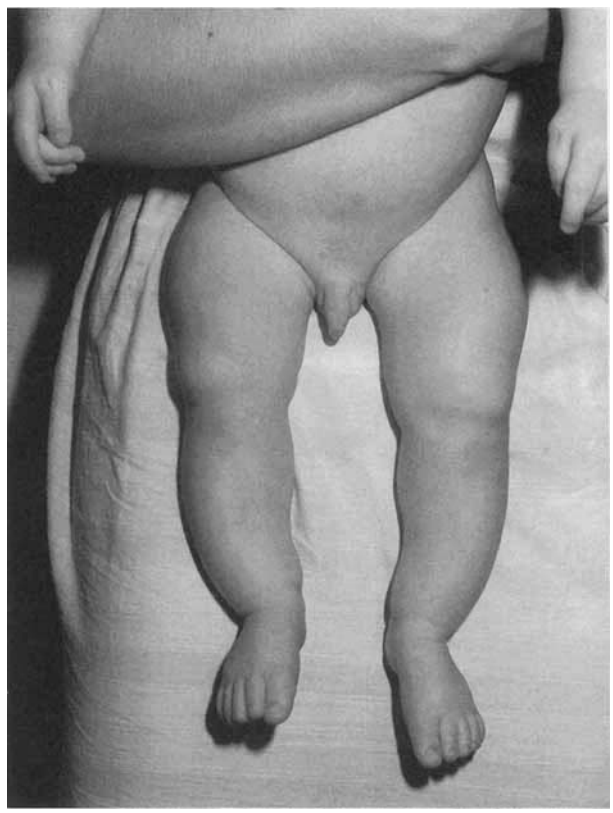

Fig. 1. One-year old. Marked bowing of legs.

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Dedicated to Jürgen W. Spranger on the occasion of his 65th birthday with admiration and best wishes.

pected. Genetic consultation was sought at 30 months. His weight was then 13 kg (25–50th centile), length was 86 cm (10th centile), and head circumference was 48 cm (−1 SD). There was some shortness of the limbs and the upper to lower segment ratio was 1.61 (normal 1.35). There was some bowing of the femora, tibiae, and radii (Fig. 1). Extension at the knees and elbows was decreased.

There was flexion at all of the small hand joints, adduction of the thumb, and camptodactyly of the 2nd–4th fingers. The fingers were short, but the palm length normal. Middle finger to hand ratio was 0.35 (3rd centile 0.38). There was talipes valgus. No cranial abnormality was seen. No limb dimples were noted. Cerebral, cardiac, abdominal, and renal ultrasound, head CT, and karyotype (QFQ banding) findings were all normal. Serum calcium, phosphate, and alkaline phosphatase were normal. Psychomotor development was slightly delayed.

X-ray examination documented bowing of the long bones of the lower limbs and of the forearms. The metaphyses were wide, of decreased transradiancy with ab-

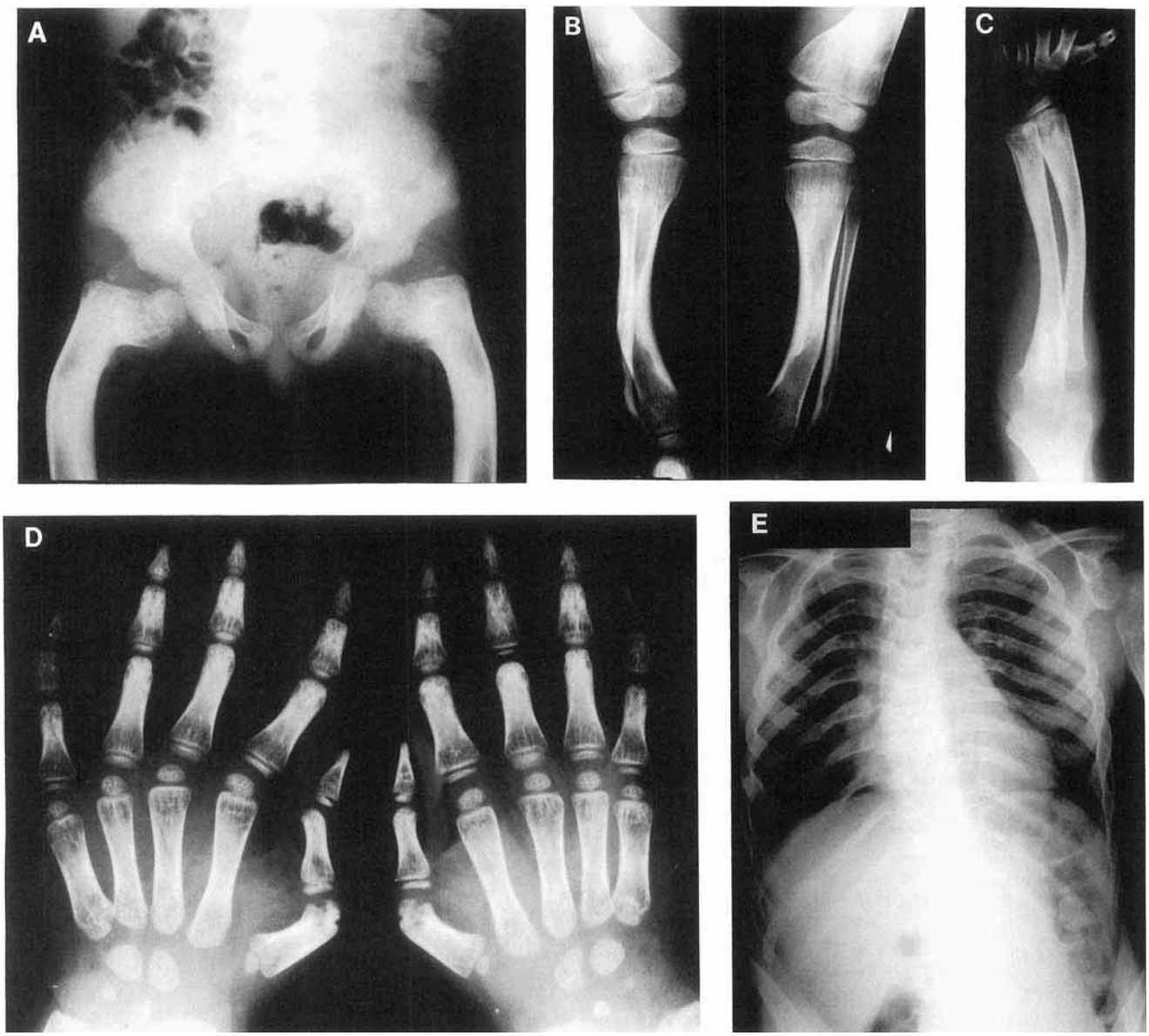

Fig. 2. **A:** Coxa vara. Short femoral necks. Note abnormal trabecular pattern of the metaphyses characterised by partly irregular longitudinal radiolucencies. Sharp bowing of the femora with internal cortical thickening at the concave site. **B:** Club-shaped metaphyses with abnormal trabecular pattern. Bowed tibiae and fibulae with internal cortical thickening. **C:** Forearm bones show similar abnormalties to those of the lower limbs. **D:** Radiolucent metaphyses with abnormal trabecular pattern. Bone age corresponds to the chronological age. **E:** The praevertebral parts of the ribs show similar changes as the metaphyses of the tubular bones. Scoliosis of the thoracic spine.

normal trabecular pattern. There was triangular cortical thickness at the site of the long bone bowing. There was bilateral coxa vara and flare of the ilia. Both the short tubular bones and the long bones showed metaphyseal radiolucency with an abnormal trabecular pattern. There was a stress fracture at the distal end of the 3rd right metatarsal. There was pinching in the posterior part of the ribs with decreased transradiancy of the posterior part. There was a mild thoracic scoliosis. The posterior and anterior borders of the vertebrae were concave; those of the upper and lower borders were convex. The interpedicular distances L1–L5 were constant. The skull was normal (Fig. 2A–E).

DISCUSSION

Stüve-Wiedemann (SW) osteochondrodysplasia is a distinct but rare and not well known disorder. Although only 2 cases were published by Stüve and Wiedemann the disorder was also observed by Maroteaux [1995] and Philippe et al. [1993]. According to Maroteaux the longest survival was 2 months. SW dysplasia has to be differentiated from two disorders: campomelic dysplasia (CD) and kyphomelic dysplasia (KD). Common clinical and radiographic finding in all is bowing of the limbs. Recurrent episodes of fever of unknown origin and feeding difficulties are common manifestations of SW dysplasia.

CD is neonatally lethal disorder with few cases surviving the first few months of life. It has a characteristic phenotype and diagnostic radiographic findings: thin long bones and hypoplastic/dysplastic scapulae and pelvis. KD is much rarer. Most patients survive and the bony deformities improve with age. Individuals have an abnormal face and skin dimples at the site of bowing. The triangular cortical thickening is not as marked as in SW dysplasia. Finally, the metaphyseal widening with decreased radiolucency and abnormal trabecular pattern is a diagnostic sign in SW dysplasia not present in CD or KD. Differential diagnosis with other long bone bowing syndromes, especially rickets and related disorders, should cause no problem: their clinical, radiographic and biochemical aspects are so different that they should not be confused with SW dysplasia [Kozlowski et al., 1978; Rezza et al., 1984; Hall and Spranger, 1980]. SW dysplasia is inherited as a recessive trait.

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